Tennessee Disorders Screened

This is a <u>screening</u> test that can be affected by baby's age, medical or treatment status at the time of specimen collection; the quality and quantity of the specimen or other variables and may not detect all affected babies. The possibility of false negative or false positive results must always be considered when screening newborns for metabolic disorders.

Disorders by MS/MS:

DISORDERS 2 Methyl 3 hydroxy butyric aciduria	2M3HBA or 2MHBD	OMIM 300256	0
2 Methylbutryl CoA Dehydrogenase Deficiency	2MBCD or 2MBG	600301	0
2,4 Dienyl CoA Reductase Deficiency	DE RED	222745	F
3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency	HMG	246450	O
3 Methyl Crotonyl CoA Carboxylase Deficiency	3MCC	210200	Ō
3 Methylglutaconyl CoA Hydratase Deficiency	3MGA	250950	О
Argininemia or Arginase Deficiency	ARG	207800	A
Argininosuccinate Lyase Def. or Argininosuccinic Aciduria	ALD or ASA	207900	A
Carnitine Palmitoyl Transferase Deficiency I	CPT I	600528	F
Carnitine Palmitoyl Transferase Deficiency II	CPT II	600650	F
Carnitine/Acylcarnitine Translocase Deficiency	CACTD	212138	F
Carnitine Uptake Deficiency	CUD	212140	F
Citrullinemia	CIT		
Type I (Arginosuccinate Synthetase Deficiency)		215700	Α
Type II (Citrin Deficiency) Glutaric Acidemia Type I	GAI	605814 231670	С
Homocystinuria	HCY	236200	A
Hypermethioninemia	HyperMet	230200	/\/
due to Glycine N-Methyltransferase Deficiency	Пурениес	606664	١.
due to S-Adenosylhomocysteine Hydrolase Deficiency		180960	A
due to Methionine Adenosyltransferase Deficiency		250850	
Hyperornithinemia	HyperOrn		
Hyperornithinemia -Hyperammonemia-Homocitrullinuria	ННН	238970	A
with Gyral Atrophy	LlumanDi-	258870	-
Hyperphenylalaninemia	HyperPhe	261600	
due to Phenylalanine Hydroxylase Deficiency due to GTP Cyclohydrolase I Deficiency		233910	
due to Pterin-4-Alpha-Carbinolamine Dehydratase Deficiency		264070	A
due to 6-Pyruvoyltetrahydropterin Synthase Deficiency		261640	"
Defects of biopterin co factor biosynthesis		261630	
Defects of biopterin co factor regeneration		182125	
Isobutyryl CoA Dehydrogenase Deficiency	IBCD	604773	С
Isovaleric Acidemia	IVA	243500	С
Long Chain Hydroxyl AcylCoA Dehydrogenase Def.	LCHAD	000000	۱_
Alpha Subunit Deficiency Beta Subunit Deficiency		600890 143450	F
Malonic Aciduria	MAL	606761	0
Maple Syrup Urine Disease	MSUD	000701	۲
Type IA	MOOD	248600	1.
Type IB		248611	A
Type II		248610	
Medium Chain AcylCoA Dehydrogenase Deficiency	MCAD	607008	F
Methylmalonic Acidemia	MMA		
due to Methylmalonyl-CoA Mutase Deficiency		251000	
due to Deficient Synthesis of 5-Prime-Deoxyadenosylcobalamin		251100	C
due to Defects in the MMAA Gene		607481	
with B12 Defect and Homocystinuria	OVT OD CKAT	277400	+
Mitochondrial Acetoacetyl CoA Thiolase Def or β Ketothiolase Multiple AcylCoA Dehydrogenase Deficiency	βKT OR SKAT MADD OR GA II	607809	С
due to Electron Transfer Flavoprotein Alpha Subunit Deficiency	WADD OR GA II	608053	
due to Electron Transfer Flavoprotein Beta Subunit Deficiency		130410	F
due to Electron Transfer Flavoprotein Dehydrogenase Deficiency		231675	
Multiple CoA Carboxylase Deficiency	MCD	253270	С
Nonketotic Hyperglycinemia	NKH		
due to Glycine Cleavage System H Protein Deficiency		238330	
due to Aminomethyltransferase Deficiency		238310	
due to Glycine Decarboxylase Deficiency	DIZLI	238300	_
Phenylketonuria Propionic Acidemia	PKU PROP	261600	A
due to Propionyl-CoA Carboxylase AlphaSubunit Deficiency	PRUP	232000	c
due to Propionyl-CoA Carboxylase Alphasubunit Deficiency		232050	
Short Chain AcylCoA Dehydrogenase Deficiency	SCAD	606885	F
Trifunctional Protein Deficiency	TFP	-	+'
Alpha Subunit Deficiency	11.5	600890	F
Beta Subunit Deficiency		143450	'
Tyrosinemia	TYR		t
Type I		276700	
		276600	A
Type II	1	276710	1
Type III		2/0/10	
	VLCAD	201475	F

Galactosemia Galt Deficiency GALT 230400

Galactokinase Deficiency Epimerase Deficiency	GALK GALE	230200 230350
Hemoglobinopathies	HGB	141800/141900
Congenital Hypothyroidism	СН	
Congenital Adrenal Hyperplasia	CAH	201910
Biotinidase Deficiency	BIOT	609019